Day 1：Nov. 7（Thu.）
Venue 1（5F 501AB）

9：10〜9：50　President Lecture

PL　Challenge to Dr. Kubota! advance

○ Mitsuru Kubota
Department of General Pediatrics & Interdisciplinary Medicine, National Center for Child Health and Development

9：50〜10：40　Oral 1：Fabry disease/Gaucher disease

Chairpersons: Ken Sakurai
（Department of Pediatrics, The Jikei University School of Medicine）
Norio Sakai
（Center for Promoting Treatment of Intractable Diseases, ISEIKAI International General Hospital）

O-1　Detection of Mulberry Bodies and Mulberry Cells by Autofluorescence using Imaging Flow Cytometry

○ Kazuya Tsuboi[1], Akinori Masago[2], Momoko Imakubo[2]
[1]LSD Center, Nagoya Central Hospital
[2]Sysmex

O-2　Chimeric anti-GLA monoclonal antibody as a reference for measuring ADA levels in Fabry patients

○ Takahiro Tsukimura[1], Daisuke Kami[2], Tomoko Shiga[3], Tadayasu Togawa[1], Satoshi Gojo[2]
Hitoshi Sakuraba[3]
[1]Department of Functional Bioanalysis, Meiji Pharmaceutical University
[2]Department of Regenerative Medicine, Graduate School of Medical Science, Kyoto Prefectural University of Medicine
[3]Department of Clinical Genetics, Meiji Pharmaceutical University

O-3　A novel mitochondrial therapy for neuropathic Gaucher Disease through cGAS-STING pathway

○ Yoshiyasu Tongu[1], Tomoko Kasahara[1], Daisuke Saigusa[2], Chikahiko Numakura[3], Kei Murayama[4]
Tomoyoshi Soga[5], Takafumi Toyohara[6], Takaaki Abe[1]
[1]Department of Clinical Biology and Hormonal Regulation, Tohoku University Graduate School of Medicine
[2]Laboratory of Biomedical and Analytical Sciences, Faculty of Pharma-Science, Teikyo University
[3]Department of Pediatrics, Yamagata University Graduate School of Medicine
[4]Diagnostics and Therapeutics of Intractable Disease, Intractable Disease Research Center and Department of Pediatrics, Juntendo University Faculty of Medicine
[5]Institute for Advanced Biosciences, Keio University
[6]Department of Nephrology, Tohoku University Graduate School of Medicine

O-4 Long term outcomes of ambroxol chaperone therapy for neuronopathic Gaucher disease: the CHANGE study

○ Aya Narita[1,2], Motomichi Kosuga[3], Torayuki Okuyama[3,4], Manabu Tanaka[5], Norio Sakai[2,6]
Chikahiko Numakura[7], Yoriko Watanabe[8], Takashi Hamazaki[9], Hiroyuki Ida[1], Kousaku Ohno[1]
[1]Department of Child Neurology, Tottori University Hospital
[2]Department of Pediatrics, ISEIKAI International General Hospital
[3]Division of Medical Genetics, National Center for Child Health and Development
[4]Department of Clinical Genomics, Saitama Medical University
[5]Division of General Pediatrics, Saitama Children's Medical Center
[6]Child Healthcare and Genetic Science Laboratory, Division of Health Sciences, Osaka University Graduate School of Medicine
[7]Department of Pediatrics, Yamagata University School of Medicine
[8]Department of Pediatrics and Child Health, Kurume University School of Medicine
[9]Department of Pediatrics, Osaka Metropolitan University Hospital
[10]Department of Pediatrics, The Jikei University School of Medicine

O-5 Phase II/III study of ambroxol hydrochloride for neuronopathic Gaucher Disease patients: J-LO study

○ Aya Narita[1,2], Shigemi Tanaka[3], Manabu Tanaka[4], Yoko Moriyama[5], Junichi Takanashi[5]
Hisashi Noma[6], Hiroshi Sunada[7], Yusuke Endo[7], Yoshihiro Maegaki[1]
[1]Department of Child Neurology, Tottori University Hospital
[2]Department of Pediatrics, ISEIKAI International General Hospital
[3]Department of pediatrics, National Hospital Organization Mie Medical Center Hospital
[4]Division of General Pediatrics, Saitama Children's Medical Center
[5]Department of pediatrics, Tokyo Women's Medical University Yachiyo Medical Center
[6]The Institute of Statistical Mathematics
[7]Advanced Medicine, Innovation and Clinical Research Center, Tottori University Hospital

10：50～11：40 Sponsored Seminar 1

Sponsored by Amicus Therapeutics K.K.

Chairperson: Norio Sakai
(Intractable Disease Center, Iseikai International General Hospital)

SPSE1 Shared Decision Making in Fabry Disease

○ Natsuko Inagaki
Department of Cardiology/Department of Clinical Genetics Center, Tokyo Medical University

12：00～12：50 Lancheon Seminar 1：Cutting Edge of PKU Management

Sponsored by BioMarin Pharmaceutical Japan K.K.

Chairperson: Kimihiko Oishi
(Department of Pediatrics, The Jikei University School of Medicine)

LS1-1 Pegvaliase Treatment During Pregnancy in Phenylketonuria Patients: A Review of Outcomes and Safety

○ Richard C. Chang
Division of Metabolic Disorders, Children's Hospital of Orange County

LS1-2 Navigating Dietary Changes in Phenylketonuria with Palynziq

○ Ilona Ginevic
Department of Genetics and Genomics, Icahn School of Medicine at Mount Sinai, New York

14：10～14：40　JSIMD Award Lecture

Chairperson: Mitsuru Kubota

(Department of General Pediatrics & Interdisciplinary Medicine, National Center for Child Health and Development)

AL　Development of a New Treatment for Inherited Mucopolysaccharidoses and Establishment of a Biomarker

○ Shunji Tomatsu
Nemours Children's Health

14：40～15：10　Encouragement Award Lecture

Chairperson: Mitsuru Kubota

(Department of General Pediatrics & Interdisciplinary Medicine, National Center for Child Health and Development)

EAL　Verification of a new screening method for inborn errors of metabolism for which newborn mass screening has not yet been established, such as late-onset OTC and CPS1 deficiency

○ Tomoko Lee
Department of Pediatrics, Hyogo Medical University

15：30～16：30　Educational Lecture 1

Chairperson: Kei Murayama

(Diagnostics and Therapeutics of Intractable Disease, Intractable Disease Center, Juntendo University Graduate School of Medicine)

EL1　Physician-Scientist Challenging Gene Therapy Innovations

○ Kazuhiro Muramatsu
Dept. of Pediatrics, Jichi Medical University

16：40～17：30　Oral 4：New treatments

Chairpersons: Kei Murayama

(Diagnostics and Therapeutics of Intractable Disease, Intractable Disease Center, Juntendo University Graduate School of Medicine)

Hiroshi Kobayashi

(Division of Gene Therapy, Research Center for Medical Science/ Department of Pediatrics/Department of Clinical Genetics, The Jikei University School of Medicine)

O-18　Phase 1/2 DTX401 Gene Therapy Study in Adults With Glycogen Storage Disease Type Ia

○ Kimimasa Tobita[1], John Michell[2], Rebecca Riba-Wolman[3], David Rodriguez-Buritica[4]
Ayesha Ahmad[5], Maria-Luz Pico[6], Terry Derks[7], David Weinstein[3], Deepali Mitragotri[8]
Andrew Grimm[8]
[1]Ultragenyx Japan K.K.
[2]Montreal Children's Hospital, Montreal, Canada
[3]University of Connecticut, Farmington, USA
[4]University of Texas McGovern Medical School, Houston, USA
[5]University of Michigan, Ann Arbor, USA
[6]Hospital Clinico Universitario de Santiago de Compostela, Santiago, Spain
[7]University of Groningen, Groningen, The Natherlands
[8]Ultragenyx Pharmaceutical Inc., Novato, USA

O-19 Therapeutic Potential of Human Amniotic Epithelial Cells in Lysosomal Storage Diseases

○ Chika Takano[1,2], Erika Ogawa[2,3], Mika Ishige[2], Isamu Taiko[4], Toshio Miki[4]
[1]Department of Pathology and Microbiology, Nihon University School of Medicine
[2]Department of Pediatrics and Child Health, Nihon University School of Medicine
[3]Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital
[4]Department of Physiology, Nihon University School of Medicine

O-20 Hereditary Coproporphyria in which the Patient's Course Improved after Givosiran Discontinuation

○ Nobuaki Ozaki, Yuri Hayashi, Jun Yoshida, Kaoru Yoshida, Atsushi Kiyota
Department of endocrinology, Japanese Red Cross Aichi Medical Center Nagoya Daiichi Hospital

O-21 Genome-editing adenovirus vector knocking-in therapeutic expression unit without cutting cell genome

○ Tomoko Nakanishi[1], Megumi Yamaji[1], Mariko Nakamura[1], Izumu Saito[2], Shigeto Sato[1]
[1]Center for Biomedical Research Resources, Juntendo University Graduate School of Medicine
[2]Department of Physiology, Juntendo University Graduate School of Medicine

O-22 A developed potent gene therapies fully rescued Neonatally Lethal Menkes disease Mouse Model

○ Miho Matakatsu[1,2], Julia Chen[3], Irene Gu[3], Jim Luo[3], Austin Gao[3], Brue Lahn[1,3]
[1]VectorBuilder Inc, Chicago, USA
[2]VectorBuilder Japan Inc
[3]Lantu biopharma（Guangzhou）Co., Ltd., Guangzhou, China